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Brief Clinical Report

Congenital Hypertrichosis, Cardiomegaly, and Osteochondrodysplasia (Cantú Syndrome): A New Case With Unusual Radiological Findings

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We report on a new case of a syndrome first described by Cantú et al. [1982: Hum Genet 60:36–41] comprising congenital hypertrichosis, "coarse" facial appearance, and mild osteochondrodysplasia. Our case has some unusual radiological findings, namely proximal and distal megaepiphyses of long bones and advanced bone age. Am. J. Med. Genet. 92:191–194, 2000. © 2000 Wiley-Liss, Inc.

KEY WORDS: generalized hyperthrichosis; osteochondrodysplasia

INTRODUCTION

In 1982, Cantú et al. described 4 patients with generalized congenital hypertrichosis, cardiomegaly, "coarse" facial appearance, and mild osteochondrodysplasia. The radiological findings were narrow thorax, broad ribs, platyspondyly, metaphyseal widening of the distal ends of long bones, bilateral coxa valga, and generalized osteopenia. Since the original report, 3 further reports were published [García-Cruz et al., 1997; Nevin et al., 1996; Rosser et al., 1998]. We report on an Italian patient presenting previously unreported radiological findings, such as proximal and distal megaepiphyses of long bones and advanced bone age.

CLINICAL REPORT

The proposita is the fourth child of a 39-year-old mother and 42-year-old father, both healthy and nonconsanguineous. She was born at 42 weeks of gestation after an uneventful pregnancy. Birth weight was 3,700 g (90–95th centile), length 51 cm (75th centile), and head circumference (OFC) 35 cm (50th centile). Growth and psychomotor devolopment have been normal.

At 3 10/12 years weight was 17.5 kg (75th centile), length 105.5 cm (75–90th centile), and head circumference 53.5 cm (>95th centile). She had a short neck, narrow thorax, and generalized hypertrichosis (Fig. 1a). The face was "coarse" with prominent eyebrows, abundant and curly eyelashes, epicanthal folds, convergent strabismus, hypertelorism, flattened and broad nasal bridge, and a long philtrum (Fig. 1b). There were no dental anomalies. Psychomotor development was normal. Laboratory investigations were normal including thyroid function tests, follicle-stimulating hormone, luteinizing hormone, estrogens, testosterone, 17 hydroxyprogesterone, urinary mucopolysaccharides and oligosaccharides, plasmatic and urinary aminoacids, and chromosomes. The computerized tomography of the brain, ophthalmological examination, and renal ultrasound were normal.

Radiographs showed a vertical base and an enlarged posterior fossa of the skull (Fig. 2a); an enlarged heart and broad ribs (Fig. 2b); hypoplastic ischio-pubic rami, bilateral coxa valga (Fig. 2c); enlarged medullary canal of the long bones with appearance of an "Erlenmeyer flask" of the distal portion of the femora (Fig. 2d); and short metacarpal and metatarsal bones with broad first metatarsals (Fig. 2e). Proximal and distal epiphyses of long bones appeared enlarged (Fig. 2d). The bone age was advanced 1 year (Greulich and Pyle Atlas and TW2 Method) [Greulich and Pyle, 1950]. There was a generalized osteopenia. The cardiological evaluation including echocardiography was normal also at 4½ years.

DISCUSSION

Congenital generalized hypertrichosis is characterized by long wavy, silky lanugo hair with only soles, palms, and mucous membranes spared. At puberty, the pubic, axillary, and beard areas retain lanugo hair and

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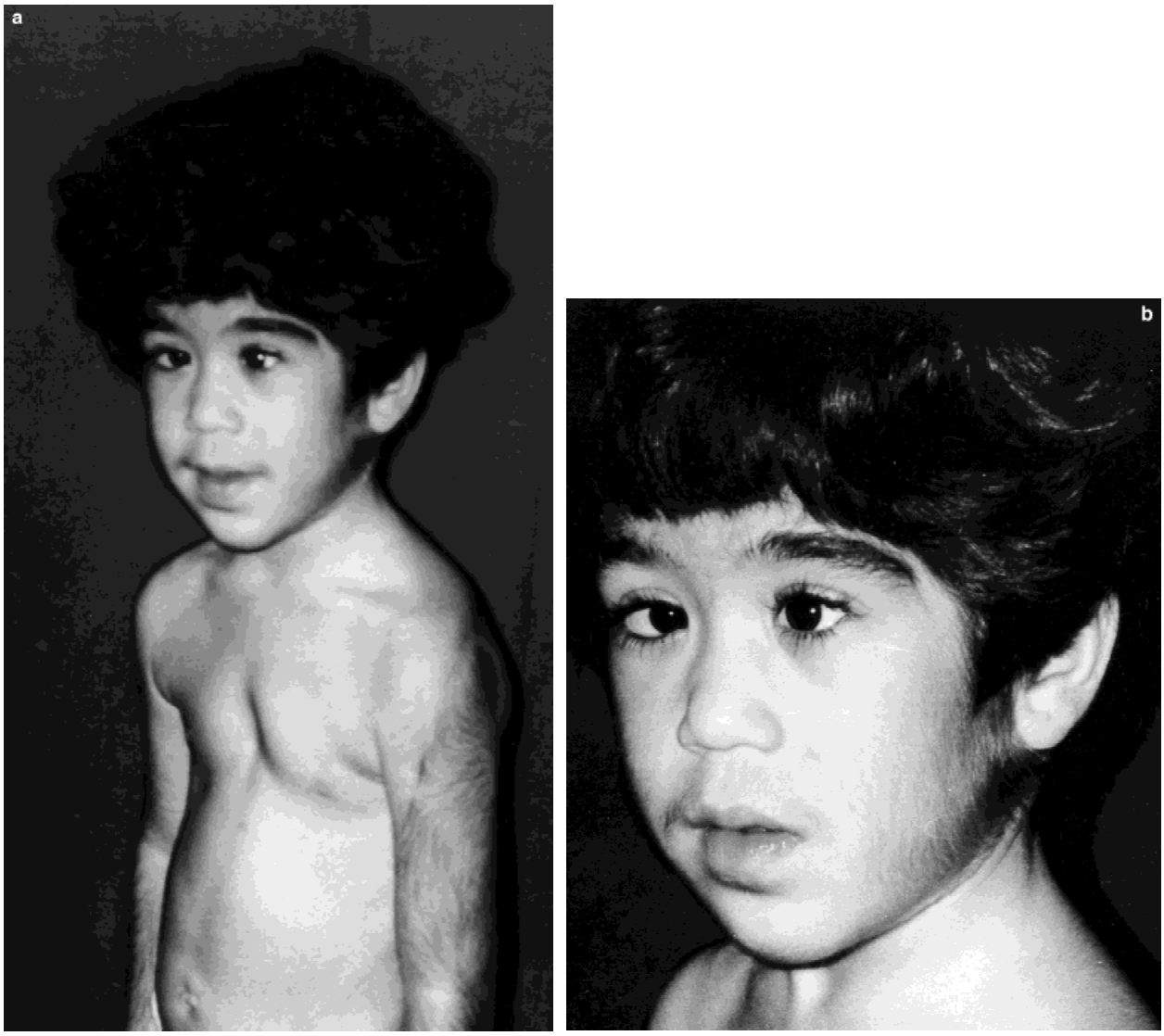

Fig. 1. The proposita at age $3^{10}/_{12}$ years: narrow thorax and generalized hypertrichosis **(a)**, and "coarse" face **(b)**.

terminal hair does not appear. This condition is a rare, fully penetrant X-linked dominant trait [Figuera et al., 1995] that may be due to a development arrest resulting in persistence of embryonic hair.

Our patient had congenital hypertrichosis universalis with "coarse" face. Several disorders with hypertrichosis and "coarse" face were considered as possible diagnoses, including hereditary hypertrichosis lanuginosa with gingival fibromatosis [Witkop, 1971], hirsutism-skeletal dysplasia-mental retardation syndrome [Wiedemann, 1993], cone-rod congenital amaurosis with congenital hypertrichosis [Jalili, 1989], and Schinzel-Giedion syndrome [Schinzel and Giedion, 1978]. However, the facial findings of prominent eyebrows, abundant and curly eyelashes, epicanthal folds, flat and broad nasal bridge, long philtrum, and a narrow thorax suggested the syndrome of congenital hypertrichosis, cardiomegaly, and osteocondrodysplasia [Cantú et al., 1982]. The radiological findings in our

TABLE I. Review of Radiological Findings in Cantú Syndrome

Radiological findings	Previous cases	Present case
Enlarged posterior fossa and vertical base of cranium	5/8	+
Broad ribs	10/12	+
Platyspondyly	7/8	–
Hypoplastic ischiopubic rami	6/7	+
Narrow obturator foramen	7/10	–
Bilateral coxa valga	7/8	+
"Erlenmeyer flask" shaped long bones and enlarged medullary canals	11/12	+
Generalized osteopenia	7/11	+
Broad first metatarsal	8/8	+
Delayed bone age	6/11	–
Advanced bone age	0/12	+
Megaepyphyses	0/12	+

Fig. 2. Radiographs: skull with vertical posterior fossa **(a)**; broad ribs, enlarged heart **(b)**; bilateral coxa valga **(c)**; enlarged medullary canal with an "Erlenmeyer-flask" appearance and proximal and distal megaepyphyses **(d)**; broad first metatarsal and short distal phalanx of the first toes **(e)**.

patient of osteopenia, wide ribs, an "Erlenmeyer flask" shaped distal portion of femora with enlarged medullary canal, and shortness of metacarpal and metatarsal bones have been described in all of the 12 previously reported patients [Cantú et al., 1982; García-Cruz et al., 1997; Nevin et al., 1996; Rosser et al., 1998]. The cardiomegaly was present in all patients; in only three cases was there associated cardiac abnormalities. Our patient did not have cardiomegaly. The clinical and radiological findings in our patient suggested a diag-

Fig. 2. (*Continued*).

nosis of Cantú syndrome. However, there were some unusual findings such as enlarged epiphises of long bones and advanced bone age, in contrast with the delayed bone age observed in other cases (Table I).

The skeletal manifestations in our case suggest variability of bone abnormalities in this syndrome. Further reports are necessary to delineate the spectrum of skeletal manifestations in this condition.

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